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Lecture

Special lecture on the care of myotonic dystrophy

—— A patient's perspective of myotonic dystrophy ——

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Last week, while preparing for my trip to Kyoto to attend the myotonic dystrophy consortium, I realized I knew nothing about the AINO Institute and Dr. Ohsawa. And so, I went to the World Wide Web and was *delighted* to read about your goals of establishing a patient-centered, medical therapeutic system. Doctors have for too long simply diagnosed and prescribed treatments, without acknowledging the whole patient. I applaud you and your goals and wish you great success in achieving them, with Dr. Ohsawa at the helm.

From my own experience, I can say that myotonic dystrophy is a little understood disease among medical professionals. It can present with a variety of symptoms, even within the same family. I would never have known I had the disease, had my older son not presented with more obvious symptoms.

I, personally, have 167 trinucleotide repeats. (You have no more than 50.) I have cataracts, a goiter, and need for a lot of sleep. I also had difficulty conceiving and delivering both of my children. I gave birth to my first son after a 36-hour labor; he spent 3 hours in the birth canal and was finally delivered with forceps. In my 2nd pregnancy, the fetus died prematurely. Then I had another son who spent his first week in ICU because of meconium aspiration.

Myotonic dystrophy is generally divided into three categories: the congenital form (the most severe form born only to affected women), childhood onset, and adult onset. My own sons suffer from childhood onset. My 25-year old son, Hunter, has 350 trinucleotide repeats. He lives with us and works as a helper in my husband's office. He is unable to wake himself up in the mornings, even with alarms and radios. He struggles with learning disabilities, inattention, socialization, myotonia, motility problems in digestion, choking, and minimal muscle wasting. He takes Adderall, an amphetamine, to keep him awake and focused. His grand-mal seizure 3 years ago and the discovery of lesions in his brain made us question the possibility of anoxia during his 36-hour birthing process. He has the classic myotonic dystrophy look with drooping eyelids, open mouth, and blunt affect. Though standardized tests reveal an IQ of 125–130, he meshes comfortably into a social group of adults with developmental disabilities. He is enthusiastic about baseball, the stock market, and has a generous spirit. He has contributed significantly to myotonic dystrophy research. His world is not always based in reality. In his last year of high school, he decided to try out for pitcher on his school baseball team, which was unrealistic, since he had not even played at all since he was 9 years old. Because he has choked and vomited several times while eating, we have learned the Heimlich maneuver. However, in his naivete and general aloofness, he does not question his diminished life. On the contrary, he is quite content.

My other son, Ashby, who is now 20, has 183 trinucleotide repeats. Though he appears to be less symptomatic than his brother and was an outstanding soccer player, psychologically he is utterly distraught because of the disease. Like his brother, he has learning disabilities and attended special schools. I also wonder if his musculature, minor skeletal abnormalities, and executive brain functioning are adversely affected. He has made many, many bad choices in his life. Just a few weeks ago, he tearfully admitted that he always felt STUPID in school, from the beginning, when he had to learn to read. He is much more socially adept than his brother, and aware of the world around him, including possible myotonic dystrophy manifestations in his children — *if he chooses* to have them.

Our journey has been long and frustrating, and I am taken back when I hear myotonic dystrophy referred to as a neuromuscular disease, because *it is so much more.* The psychological and emotional aspects of myotonic dystrophy have plagued our family, thus far, more significantly than the physical ones. We searched for seven years to find a correct diagnosis, beginning with a highly renowned pediatric neurologist when Hunter was 6, who laughed at us and assured us nothing was wrong; so we continued to address specific symptoms as they arose. When he was 12, a psychiatrist diagnosed him as a childhood schizophrenic. Another neurologist finally diagnosed him correctly in ten minutes. Because I have found few doctors in Atlanta who truly understand the disease, for years, I carried Prof. Harper's book on myotonic dystrophy to doctors' appointments. As a result of Hunter's diagnosis, our pediatrician started including the finger squeeze to check for myotonia in his annual physicals for *all* patients.

Some of the most challenging aspects of myotonic dystrophy, which impact every facet of the patient's life, are general *apathy* and *inertia.* Dr. Peter Harper writes in his book on myotonic dystrophy: "We have found that affected individuals, when just mildly incapacitated, were often content to sit or lie idly for hours."

Dr. Harper also describes the *unkempt nature* of many myotonic dystrophy patients:

> "While in the country in search of a certain "myotonic's" home, it was often possible to identify a residence by its neglected appearance, the obvious need of repairs, the unkempt yard and garden choked with overgrown grass and weeds, which provided a vivid contrast with the surrounding well-kept homes."
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> (Harper, p. 153)

Many don't mind living with complete disorder, or even squalor.

My own sons are content to watch TV or play Nintendo and computer games for endless hours. Their choice of entertainment has been a source of discord in our family. As a parent, I have struggled with the question, "What are appropriate expectations for them?" With Hunter, I finally came to have NO expectations; now, any time he does anything well, we celebrate! With Ashby, we still do not know what appropriate expectations should be for him. We as parents are constantly questioning when our "encouragement and support" are perceived as "control and pressure."

Since Hunter was diagnosed 12 years ago, I have discovered that I have an insatiable curiosity and a lot of passion about things that matter to me. And, I believe that knowledge is power. After several years of research and living with the disease, I reached a personal, *ethical dilemma.* I had an enormous amount of information, and although the disease had affected the Hunter branch of my family for years, no one seemed to know its correct name. It was affectionately referred to as the "Hunter disease." I realized that if someone had known what I know now, when I was *planning* my family, and failed to inform me, that I would be *angry.* And so, in February of 1997, I wrote a 4-page letter to my generation of relatives and older. I informed them about this genetic disease and how and where they could pursue DNA testing *if they chose.* I suggested they share it with their children *if/when* they saw fit. About 65% contacted me and thanked me for the information, the other 35% did not. I have learned it is difficult for people to discuss a devastating genetic disorder. Only 1 out of 4 of my siblings has had DNA testing, and like me, he has the disorder. Having no role models to emulate, ethically, I did what I my inner voice compelled me to do when I mailed the letter to my family.

I observe members of my extended family with apathy, low motivation, cataracts, a tremendous need for sleep, and many other symptoms. My younger brother has fallen asleep at the table during dinner parties. I have a 5-year old cousin with the congenital form, who could not breathe or swallow at birth. But because of medical heroics, she lived. At age 5, she has finally learned to crawl across the floor to retrieve her bottle. She has had eye, leg, and heart surgery. She wears leg braces and can utter only one sound: *daaaa.* She goes to physical, occupational and speech therapy every week.

And so... what does one do when faced with such a diagnosis?

My life will be forever divided into *life before and life after diagnosis.* After going through a dark, depressed period, I finally emerged with more purpose, focus, and passion than before, and with the realization that I was a mortal being and must live each day fully. Several factors helped my mind and

spirit arrive at a happier place: 1) *deep human connection*, through therapy and support groups; 2) a *sense of control over my attitude*, which I finally discovered in the process; 3) the *appreciation and creation of art*; 4) and a *sense of humor.*

Now, what would *my advice*, as a myotonic dystrophy patient, be to you nursing students who will be treating people with this disease? I have 10 suggestions:

1) Remember that in spite of their open-mouths, drooping eyelids, garbled speech, they are human beings experiencing the same emotions as everyone else. *Spend time with them, and listen well.* Respond to them with kindness and patience. *What every human being yearns for is an honest response from another human being.* Giving them your time honors and validates them.
2) Encourage them to *participate in organized activities*, planned by someone other than themselves. Sometime they lack the ability to initiate but can respond to someone else's encouragement.
3) *Offer group therapy*, under trained leadership, where all emotions can be shared and explored. Life is difficult to manage alone. Somehow, in sharing one's stories in the face of others who listen well, the negative emotions magically start to diminish.
4) *Encourage them to eat well*, a balanced diet with adequate roughage for proper digestion. This is very important.
5) *Create opportunities* for moderate *physical exercise.*
6) *Wake them up* in the morning if that's what it takes.
7) *Incorporate the appreciation and creation of art* in their lives.
8) *Give them small responsibilities*, which will help them value themselves.
9) *Tailor educational environments to meet their special needs.*
10) And of course, *proper medical attention* goes without saying. Since we have no treatment yet, we can only deal with specific symptoms as they arise.

Also, remember that we all need *someone to love, something to do, and something to look forward to.* It sounds simplistic, but it's true. Fortunately, I have a number of people in my life to love; and I realized a long time ago, I had to depend on people like Drs. Ashizawa and Ohsawa to try find a treatment for MYOTONIC DYSTROPHY, that I lacked the expertise. But I could raise money for myotonic dystrophy research. And something I have looked forward to is my trip to Japan. When I am feeling depressed, I try to plan something fun — lunch with a friend — even something as simple as taking a warm bath.

For me, the quality of one's life is so much more important than its length. YOU will be in a position to enhance the patients' lives; you can give them *hope.*

But in the process, above all, *take care of yourself.* Your well of giving is not bottomless. You can give until you give out if you don't nourish your own spirit along the way. Flight attendants instruct mothers in case of emergency to put the oxygen mask on themselves, and then on the children. Remember to keep yourself healthy and strong so that you can continue to care for your patients.

Thank you for the opportunity to be with you today.